SCN2A
AUSTRALIA

INFORMATION SHEET

What is SCN2A?

A plain-language guide for families

SCN2A is a gene, one of the sets of instructions your body uses to grow and work. The SCN2A gene tells the brain how to build a tiny protein called a **sodium channel** (named $Na_V1.2$) that helps brain cells send signals to each other.[1,3] When there is a change in SCN2A, called a **variant**, those signals can fire too much or too little. That can affect how a child has seizures, learns, moves and develops.

THE KEY POINTS

- A change in SCN2A can cause a range of conditions, from mild to severe.[2]
- Common features are epilepsy (seizures), developmental delay or intellectual disability, autism, and differences in movement.[2]
- Most changes are new in the child (called **de novo**) and are not inherited from a parent.[1,2]
- There is no SCN2A-specific cure yet, but a great deal can be done to manage symptoms, and targeted treatments are in development.
- The *type* of change matters: it can guide which seizure medicines are likely to help.[1,3]

Two broad types of SCN2A change

Most SCN2A variants act in one of two ways. Knowing which one your child has is one of the most useful things a genetic result can tell you.[1,3]

CHANNEL TOO ACTIVE

Gain-of-function

Seizures usually start **early, often before three months** of age. These children frequently respond well to a group of medicines called **sodium channel blockers** (for example phenytoin, carbamazepine or oxcarbazepine).[1]

CHANNEL UNDERACTIVE

Loss-of-function

Seizures, if they occur, tend to start **later (after three months)**. Some children have autism or intellectual disability with few or no early seizures. Sodium channel blockers often don't help, and can sometimes make seizures worse.[1,3]

Worth asking your team: “Is my child's variant gain-of-function or loss-of-function, and what does that mean for which seizure medicines we try?”

How common is it?

SCN2A-related conditions are rare. Current estimates put them at around **1 in 80,000 births**, and that is probably an undercount, because not everyone is tested.[2] Even so, SCN2A is one of the more common single-gene causes of severe early epilepsy, behind about 1 in 100 cases of epileptic encephalopathy.[2]

What helps

- A **paediatric neurologist** for seizures, and a paediatrician to coordinate care.
- **Early therapies** (speech, occupational therapy, physiotherapy) to support development.
- A **genetics service** to explain your child's specific variant and what it means for your family.
- **Other families**, through SCN2A Australia, for connection and lived-experience knowledge.

WORDS YOU MIGHT HEAR

- **Variant** — a change in the gene.
- **De novo** — new in the child, not inherited.
- **Gain- / loss-of-function** — whether the channel is too active or underactive.
- **Sodium channel blocker** — a type of seizure medicine that helps some, but not all, children.

SOURCES

1. Wolff M, et al. Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. *Brain*. 2017;140(5):1316–1336.
2. Wolff M, et al. Phenotypic spectrum and genetics of SCN2A-related disorders, treatment options and outcomes. *Epilepsia*. 2019;60(S3):S59–S67.
3. Sanders SJ, et al. Progress in understanding and treating SCN2A-mediated disorders. *Trends Neurosci*. 2018;41(7):442–456.

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